Appendix A: Denominators Used in Calculating Rates

Year	Breakdown		Live births
1998* and 1999 Combined	Overall		641,337
	By Mother's Age	Less than 20	102,851
		20 to 24	180,265
		25 to 29	173,432
		30 to 34	120,271
		35 to 39	54,085
		40 +	10,343
	By Mother's Race/Ethnicity	White	258,135
		African American	71,018
		Hispanic	292,141
		Other	19,293
	By Infant's Sex	Male	327,711
		Female	313,626
	By Region	Region 01	24,324
		Region 02	14,888
		Region 03	180,401
		Region 04*	20,297
		Region 05*	15,217
		Region 06*	124,323
		Region 07	70,845
		Region 08	69,149
		Region 09	16,501
		Region 10	29,134
		Region 11	76,258
1996†	Overall		114,084
1997‡	Overall		186,347
1998*	Overall		292,180
1999	Overall		349,157

* For three Regions, 1998 data are limited to half of the year: January 1-June 30, 1998, in Region 04; July 1-December 31, 1998, in Regions 05 and 06.

† In 1996, the registry covered Regions 6 and 11 only.

‡ In 1997, the registry covered Regions 2, 3, 8, 9, 10, and 11.

Appendix B: BPA Codes Used to Define Birth Defects Shown in this Report

Diagnoses in the Texas Birth Defects Registry are coded using a system developed and provided by the National Center for Environmental Health of the Centers for Disease Control and Prevention (CDC). The birth defect codes, commonly called BPA codes, are based on the British Pediatric Association Classification of Diseases (1979) and the World Health Organization's International Classification of Diseases, 9th Revision, Clinical Modification (ICD-9-CM) (1979).

The table below shows the BPA codes used to define the conditions shown in this report.

Birth defect	BPA codes
Agenesis, aplasia, or hypoplasia of the lung	748.500 - 748.510
Anencephaly	740.000 - 740.100
Aniridia	743.420
Anophthalmia	743.000
Anotia or microtia	744.010, 744.210
Aortic valve stenosis	746.300
Atrial septal defect	745.510 - 745.590
Biliary atresia	751.650
Bladder exstrophy	753.500
Cataract	743.320, 743.325, 743.326
Choanal atresia or stenosis	748.000
Cleft lip with or without cleft palate	749.100 - 749.220
Cleft palate alone (without cleft lip)	749.000 - 749.090
Coarctation of the aorta	747.100 - 747.190
Common truncus	745.000 - 745.010
Congenital hip dislocation	754.300
Craniosynostosis	756.000 - 756.030
Diaphragmatic hernia	756.610 - 756.617
Ebstein anomaly	746.200
Encephalocele	742.000 - 742.090
Endocardial cushion defect	745.600 - 745.690
Fetal alcohol syndrome or other alcohol related birth defects	760.710, 760.720
Gastroschisis	756.710
Hirschsprung disease	751.300 - 751.340
Holoprosencephaly	742.260
Hydrocephaly	742.300 - 742.390, excluding 742.385
Hypoplastic left heart syndrome	746.700
Hypospadias or epispadias	752.600 - 752.627, excluding 752.621

Birth defect	BPA codes
Microcephaly	742.100
Microphthalmia	743.100
Obstructive genitourinary defect	753.200 - 753.290 and 753.600 - 753.690
Omphalocele	756.700
Patent ductus arteriosus	747.000
Pulmonary valve atresia or stenosis	746.000 - 746.010
Pyloric stenosis	750.510
Reduction defects of the lower limbs	755.300 - 755.390
Reduction defects of the upper limbs	755.200 - 755.290
Renal agenesis or dysgenesis	753.000 - 753.010
Spina bifida without anencephaly	741.000 - 741.990, without 740.000 - 740.100
Stenosis or atresia of large intestine, rectum, or anal canal	751.200 - 751.240
Stenosis or atresia of small intestine	751.100 - 751.195
Tetralogy of Fallot	745.200 - 745.210, 746.840
Tracheoesophageal fistula / esophageal atresia	750.300 - 750.350
Transposition of the great vessels	745.100 - 745.190
Tricuspid valve atresia or stenosis	746.100
Trisomy 13 (Patau syndrome)	758.100 - 758.190
Trisomy 18 (Edwards syndrome)	758.200 - 758.290
Trisomy 21 (Down syndrome)	758.000 - 758.090
Ventricular septal defect	745.400 - 745.490

Appendix C: Glossary of Birth Defect Terms

Agenesis Absence of part(s) of the body.

Agenesis, aplasia, or hypoplasia of the lung The absence or incomplete development of a lung or lung tissue.

Anencephaly Congenital absence of the skull, with cerebral hemispheres completely missing or reduced to small masses attached to the base of the skull. Anencephaly is not compatible with life.

Aniridia The complete absence of the iris of the eye or a defect of the iris. Can be congenital or traumatically induced.

Anopthalmia A developmental defect characterized by complete absence of the eyes, or by the presence of vestigial eyes.

Anotia A congenital absence of one or both ears.

Aortic valve stenosis A cardiac anomaly characterized by a narrowing or stricture of the aortic valve. This condition causes abnormal cardiac circulation and pressure in the heart during contractions. This condition can be repaired surgically in some cases.

Atresia Imperforation; absence or closure of a normal opening.

Atrial septal defect A congenital cardiac malformation in which there are one or several openings in the atrial septum (muscular and fibrous wall between the right and left atria) allowing a mixing of oxygenated and unoxygenated blood. The openings vary in size and may resolve without treatment or may require surgical treatment. Also called *ostium secundum defect.*

Biliary atresia A congenital absence or underdevelopment of one or more of the ducts in the biliary tract. Correctable surgically.

Bladder exstrophy Incomplete closure of the anterior wall of the bladder and the abdominal cavity. The upper urinary tract is generally normal. Often associated with anorectal and genital malformations, and epispadias. Affected persons are at a markedly increased risk of bladder carcinoma (squamous cell). This condition is usually corrected surgically after birth.

Cataract An opacity (clouding) of the lens of the eye.

Choanal atresia or stenosis A congenital anomaly in which a bony or membranous formation blocks the passageway between the nose and the pharynx. This defect is usually repaired surgically after birth. Bilateral choanal atresia is a surgical emergency.

Cleft lip The congenital failure of the fetal components of the lip to fuse or join, forming a groove or fissure in the lip. Infants with this condition can have difficulty feeding, and may use assistive devices for feeding. This condition is corrected when the infant can tolerate surgery.

Cleft palate The congenital failure of the palate to fuse properly, forming a grooved depression or fissure in the roof of the mouth. This defect varies in degree of severity. The fissure can extend into the hard and soft palate and into the nasal cavities. Infants with this condition have difficulty feeding, and may use assistive devices for feeding. Surgical correction is begun as soon as possible. Children with cleft palates are at high risk for hearing problems due to ear infections.

Coarctation of the aorta Localized narrowing of the aorta. This condition causes abnormal cardiac circulation and pressure in the heart during contractions. This condition can vary from mild to severe. Surgical correction is recommended even for mild defects.

Common truncus ateriosus A congenital heart defect in which the common arterial trunk fails to divide into pulmonary artery and aorta. This is corrected surgically.

Confidence interval (95%) The interval that contains the true prevalence (which we can only estimate) 95% of the time. See Methods for more explanation.

Congenital Existing at or dating from birth.

Congenital hip dislocation A congenital defect in which the head of the femur does not

articulate with the acetabulum of the pelvis because of an abnormal shallowness of the acetabulum. Treatment in early infancy consists of bracing of the joint to cause a deepening of the acetabulum.

Craniosynostosis A premature ossification (closing) of the cranial sutures before birth or soon after birth. This condition is occasionally associated with other skeletal defects. If no surgical correction is made, the growth of the skull is inhibited, and the head is deformed. The eyes and the brain are often damaged.

Diaphragmatic hernia A failure of the diaphragm to form completely, leaving a hole. Abdominal organs can protrude through the hole into the chest cavity and interfere with development of the heart and lungs. Usually life-threatening and requires emergent surgery.

Down syndrome (Trisomy 21) The chromosomal abnormality characterized by an extra copy of chromosome 21. In rare cases this syndrome is caused by translocation. The extra copy can be free-lying, or can be attached to some other chromosome, most frequently number 14. Down syndrome can occur in mosaic, so that there is a population of normal cells and a population of trisomy 21 cells. Down syndrome is characterized by moderate to severe mental retardation, sloping forehead, small ear canals, flat bridged nose, and short fingers and toes. One third of infants have congenital heart disease, and one third have duodenal atresia. (Both can be present in the same infant.) Affected people can survive to middle or old age. There is an increased incidence of Alzheimer disease in adults with Down syndrome.

Ebstein anomaly A congenital heart defect in which the tricuspid valve is displaced downward into the right ventricle causing abnormal patterns of cardiac circulation.

Edwards syndrome (Trisomy 18) The chromosomal abnormality characterized by an extra copy of chromosome 18. The extra chromosome can be free lying or attached to another chromosome. Trisomy 18 can occur in mosaic. Edwards syndrome is characterized by mental retardation, neonatal hepatitis, low-set ears, skull malformation, and short digits. Cardiac and renal anomalies are also common. Survival for more than a few months is rare.

Encephalocele The protrusion of the brain substance through a defect in the skull.

Endocardial cushion defect A variety of septal defects (malformations of the walls separating the two atria and two ventricles of the heart) resulting from imperfect fusion of the endocardial cushions in the embryonic heart.

Epispadias A congenital defect in which the urinary meatus (urinary outlet) opens above (dorsal to) the normal position. The urinary sphincters are defective, so incontinence does occur. Surgical correction is aimed at correcting incontinence and permitting sexual functioning. The corresponding defect in females is rare. *See also Hypospadias.*

Esophageal stenosis or atresia A narrowing or incomplete formation of the esophagus. Usually a surgical emergency. Frequently associated with a tracheoesophageal fistula.

Fetal alcohol syndrome A constellation of physical abnormalities (including characteristic abnormal facial features and growth retardation), and problems of behavior and cognition in children born to mothers who drank alcohol during pregnancy.

Fistula An abnormal passage from an internal organ to the body surface or between two internal organs or structures.

Gastroschisis A congenital opening of the abdominal wall with protrusion of the intestines. This condition is surgically treated. Contrast with Omphalocele, below.

Hirschsprung disease The congenital absence of autonomic ganglia (nerves controlling involuntary and reflexive movement) in the muscles of the colon. This results in immobility of the intestines and may cause obstruction or stretching of the intestines. This condition is repaired surgically in early childhood by the removal of the affected portion of the intestine.

Holoprosencephaly Failure of the brain to develop into two equal halves, so there is structural abnormality of the brain. There may be associated midline facial defects including cyclopia (fusion of the eye orbits into a single cavity containing one eye) in severe cases. About half the cases are probably due to a single gene

defect (the HPE gene). Frequently occurs with Trisomy 13.

Hydrocephaly The abnormal accumulation of fluid within the spaces of the brain.

Hyperplasia Overgrowth characterize by an increase in the number of cells of a tissue.

Hypoplasia A condition of arrested development in which an organ or part remains below the normal size or in an immature state.

Hypoplastic left heart syndrome Atresia, or marked hypoplasia, of the aortic opening or valve, with hypoplasia of the ascending aorta and defective development of the left ventricle (with mitral valve atresia). This condition can be surgically repaired in a series of three procedures over a period of one year. Transplantation is also a treatment. This condition is usually fatal in the first month of life if not treated.

Hypospadias A congenital defect in which the urinary meatus (urinary outlet) is on the underside of the penis or on the perineum (area between the genitals and the anus). The urinary sphincters are not defective so incontinence does not occur. The condition may be surgically corrected if needed for cosmetic, urologic, or reproductive reasons. The corresponding defect in women is rare. *See also epispadias*

Limb defects See Reduction defects.

Meninges Membranes that cover the brain and spinal cord.

Microcephaly The congenital smallness of the head, with corresponding smallness of the brain.

Microphthalmia The congenital abnormal smallness of one or both eyes. Can occur in the presence of other ocular defects.

Microtia A small or maldeveloped external ear and atretic or stenotic external auditory canal.

Mosaic In genetics, this refers to an individual organism that has two or more kinds of genetically different cell types. The degree of abnormality depends on the type of tissue containing affected cells. Individuals may vary from near normal to full manifestation of the genetic syndrome. Can occur in any chromosome abnormality syndrome.

Neural tube defect A defect resulting from failure of the neural tube to close in the first month of pregnancy. The major conditions include anencephaly, spina bifida, and encephalocele.

Obstructive genitourinary defect Stenosis or atresia of the urinary tract at any level. Severity of the defect depends largely upon the level of the obstruction. Urine accumulates behind the obstruction and damages the organs.

Omphalocele The protrusion of an organ into the umbilicus. The defect is usually closed surgically soon after birth. Contrast with Gastroschisis.

Patau syndrome (Trisomy 13) The chromosomal abnormality caused by an extra chromosome 13. The extra copy can be free-lying, or can be attached to some other chromosome. Patau syndrome can occur in mosaic so that there is a population of normal cells and a population of trisomy 13 cells. Patau syndrome is characterized by impaired midline facial development, cleft lip and palate, polydactyly, and mental retardation. Most infants do not survive beyond 6 months of life.

Patent ductus arteriosus A blood vessel between the pulmonary artery and the aorta. This is normal in fetal life, but can cause problems after birth, particularly in premature infants. This condition causes abnormal cardiac circulation and pressure in the heart during contractions. The vast majority close spontaneously and cause no problems. Medical or surgical correction may be done. This is only an abnormality if it causes significant medical problems.

Pulmonary artery anomaly Abnormality in the formation of the pulmonary artery such as stenosis or atresia. See also common truncus.

Pulmonary valve atresia or stenosis A congenital heart condition characterized by absence or constriction of the pulmonary valve. This condition causes abnormal cardiac circulation and pressure in the heart during contractions. This condition can vary from mild to severe. Mild forms are relatively well

tolerated and require no intervention. More severe forms are surgically corrected.

Pyloric stenosis A narrowing of the pyloric sphincter at the outlet of the stomach. This causes a blockage of food from the stomach into the small intestine. Usually treated surgically.

Reduction defects of the lower limbs The congenital absence of a portion of the lower limb. There are two general types of defect, transverse and longitudinal. Transverse defects appear like amputations, or like missing segments of the limb. Longitudinal defects are missing rays of the limb (for example, a missing tibia and great toe).

Reduction defects of the upper limbs The congenital absence of a portion of the upper limb. There are two general types of defect, transverse and longitudinal. Transverse defects appear like amputations, or like missing segments of the limb. Longitudinal defects are missing rays of the limb (for example, a missing radius and thumb).

Renal agenesis or dysgenesis The failure, or deviation, of embryonic development of the kidney.

Spina bifida A neural tube defect resulting from failure of the spinal neural tube to close. The spinal cord and/or meninges may or may not protrude. This usually results in damage to the spinal cord with paralysis of the involved limbs. Includes myelomeningocele (involving both spinal cord and meninges) and meningocele (involving just the meninges).

Stenosis A narrowing or constriction of the diameter of a bodily passage or orifice.

Stenosis or atresia of large intestine, rectum and anus The absence, closure or constriction of the large intestine, rectum or anus. Can be surgically corrected or bypassed.

Stenosis or atresia of the small intestine A narrowing or incomplete formation of the small intestine obstructing movement of food through the digestive tract.

Tetralogy of Fallot A congenital cardiac anomaly consisting of four defects: ventricular septal defect, pulmonary valve stenosis or atresia, displacement of the aorta to the right, and hypertrophy of right ventricle. The condition is corrected surgically.

Tracheoesophageal fistula An abnormal passage between the esophagus and trachea. Leads to pneumonia. Corrected surgically. It is frequently associated with esophageal atresia.

Translocation The rearrangement of genetic material within the same chromosome or the transfer of a segment of one chromosome to another one. People with balanced translocations do not always manifest genetic syndromes, but may be carriers of genetic syndromes and can have children with unbalanced translocations. Can occur with any chromosomal anomaly syndrome.

Transposition of the great vessels A congenital malformation in which the aorta arises from the right ventricle and the pulmonary artery from the left ventricle (opposite of normal), so that the venous return from the peripheral circulation is recirculated without being oxygenated in the lungs. Immediate surgical correction is needed. When this is not associated with other cardiac defects, and not corrected, it is fatal.

Tricuspid valve atresia or stenosis A congenital cardiac condition characterized by the absence or constriction of the tricuspid valve. The opening between the right atrium and right ventricle is absent or restricted, and normal circulation is not possible. This condition is often associated with other cardiac defects. This condition is surgically corrected depending on the severity.

Trisomy A chromosomal abnormality characterized by one more than the normal number of chromosomes. Normally, cells contain two of each chromosome. In trisomy, cells contain three copies of a specific chromosome.

Trisomy 13 (Patau syndrome) The chromosomal abnormality caused by an extra chromosome 13. The extra copy can be free-lying, or can be attached to some other chromosome. Trisomy 13 can occur in mosaic so that there is a population of normal cells and a population of trisomy 13 cells. Trisomy 13 is characterized by impaired midline facial development, cleft lip and palate, polydactyly, and mental retardation. Most infants do not survive beyond 6 months of life.

Trisomy 18 (Edwards Syndrome) The chromosomal abnormality characterized by an extra copy of chromosome 18. The extra chromosome can be free lying or attached to another chromosome. Trisomy 18 can occur in mosaic so that there is a population of normal cells and a population of trisomy 18 cells. Trisomy 18 is characterized by mental retardation, neonatal hepatitis, low-set ears, skull malformation, and short digits. Cardiac and renal anomalies are also common. Survival for more than a few months is rare.

Trisomy 21 (Down Syndrome) The chromosomal abnormality characterized by an extra copy of chromosome 21. In rare cases this syndrome is caused by translocation. The extra copy can be free-lying, or can be attached to some other chromosome, most frequently number 14. Trisomy 21 can occur in mosaic, so that there is a population of normal cells and a population of trisomy 21 cells. Trisomy 21 is characterized by moderate to severe mental retardation, sloping forehead, small ear canals, flat bridged nose, and short fingers and toes. One third of infants have congenital heart disease, and one third have duodenal atresia. (Both can be present in the same infant.) Affected people can survive to middle or old age. There is an increased incidence of Alzheimer disease in adults with Trisomy 21.

Truncus arteriosus *See Common truncus*.

Ventricular septal defect (VSD) A congenital cardiac malformation in which there are one or several openings in the ventricular septum (muscular and fibrous wall between the right and left ventricle or right and left lower chambers of the heart) allowing a mixing of oxygenated and unoxygenated blood. The openings vary in size and may resolve without treatment or require surgical treatment.

Appendix D: Birth Defect Investigations, 1998 and 1999

In addition to routine data collection, we conduct birth defect investigations throughout the state. Health care professionals and the public can report apparently unusual concentrations (also called "clusters") of birth defects to the Texas Birth Defects Monitoring Division. Birth defect investigations are then initiated to determine if there is a statistically significant elevation in the number of children with birth defects. In 1998 and 1999, 26 birth defect clusters were investigated, as shown below.

Condition	Geographic area
Anencephaly	Bastrop County
Hypoplastic left heart syndrome	Brazos County
Anencephaly	Bryan and College Station
Neural tube defects	Cameron County
Neural tube defects	Collin and Denton Counties
Anencephaly	Conroe
Neural tube defects	Dallas
Biliary atresia	El Paso
Multiple birth defects	El Paso
Neural tube defects	El Paso
Trisomy 18	El Paso
Multiple birth defects	Ellis County
Cleft lip and cleft palate	Gaines and Dawson Counties
Multiple birth defects	Grand Prairie
Multiple birth defects	Kelly Air Force Base (Bexar County)
Neural tube defects	Laredo
Anencephaly and Holoprosencephaly	Nueces County
Cleft lip and cleft palate	Ozona
Gastroschisis	Port Lavaca
Cleft lip and cleft palate	Presidio and Alpine
Multiple birth defects	Roby
Heart defects	San Angelo and Ballinger
Down syndrome	Smith County
Agenesis of the corpus callosum	Tom Green County
Omphalocele and Gastroschisis	Travis County
Birth defects (eye defects)	West Dallas

More detailed reports of the birth defect investigations conducted in 1998 and 1999 can be obtained by visiting on our web site at www.tdh.state.tx.us/tbdmd/index.htm or by contacting the Texas Birth Defects Monitoring Division at (512) 458-7232.

Appendix E: Research Using Data from the Texas Birth Defects Registry and Other Related Projects

Once in the Texas Birth Defects Registry, a case infant's family may be invited to participate in research studies. Epidemiologists and clinicians conduct epidemiological research to investigate the causes of birth defects. Members of the Texas Department of Health Institutional Review Board annually evaluate the protocols for each study to protect the privacy and other concerns of participants.

Below are descriptions of research using data contained in the Texas Birth Defects Registry and other projects related to the registry or birth defects. In addition to the publications listed below, data are occasionally published as Special Reports in the Texas Birth Defects Monitor, the twice-yearly newsletter of the Texas Birth Defects Monitoring Division. Past issues of the Monitor are available on our website at www.tdh.state.tx.us/tbdmd/monitor/the.htm.

Published Research Articles using Registry Data

Canfield MA, Anderson JL, Waller DK, Palmer S, Kaye C. 2002. **Folic acid awareness and use among women with a history of a neural tube defect pregnancy, Texas, 2000-2001**. MMWR 51(No. RR-13):16-19. **Abstract:** The use of folic acid is a critical component in preventing birth defects. Health-care providers should take advantage of all health-care visits to counsel not only women at high risk (i.e., those with a history of having an infant with a neural tube defect [NTD]) but all women regarding the importance of folic acid use. A study conducted in Texas confirmed that white and Hispanic mothers were equally likely to recall receiving postpartum advice to use folic acid; however, Hispanic women were much less likely to use folic acid, compared with white women. This report covers data from May 2000 through November 2001. A study was conducted in Texas to determine whether women at high risk recall and follow recommendations to use folic acid. The study included 195 women at high risk and 223 control mothers who gave birth to infants without birth defects. These women participated in a telephone interview for a population-based case-control study of NTDs. Approximately 56.4% (110 of 195) of mothers who had infants affected by an NTD recalled receiving postpartum advice to use folic acid, compared with 25.6% (57 of 223) of control mothers ($p < 0.01$). Among nonpregnant case mothers, 54 (32.7%) of 165 reported regular use of supplements containing folic acid, and 53 (25.2%) of 210 nonpregnant control mothers reported this behavior ($p = 0.11$). Among case mothers, use of folic acid was significantly higher for whites (64.7%) versus Hispanics (16.5%) ($p < 0.001$); for women with some college education (57.1%) versus no college education (20.2%; $p < 0.001$); for women who were trying to get pregnant (66.7%) versus those using birth control (38.3%) or reporting using no contraceptive method (18.8%) ($p = 0.001$); and for women who reported receiving advice to use folic acid (40.9%) versus those who did not (22.2%; $p = 0.01$). Findings from this study support the need to implement NTD recurrence prevention activities in Texas. Data also identify a need for educational strategies in Texas that target Hispanic women at high risk, especially those who primarily speak Spanish. Further efforts should be made to determine why Hispanic women have low rates of folic acid use (e.g., the cost of vitamins and language and cultural barriers). On the basis of a review of research and current practice, recommendations developed by the Public Health Service include 1) women at risk for a recurrent NTD-affected pregnancy should take 0.4 mg of folic acid per day; and 2) if a woman at high risk is planning a pregnancy, she should consult her physician regarding taking the higher dose of 4.0 mg per day.

Ethen MK, Canfield MA. 2002. **Impact of including elective pregnancy terminations before 20 weeks gestation on birth defect rates**. Teratology 66:S32-S35. This material is used by permission of Wiley-Liss, Inc., a subsidiary of John Wiley & Sons, Inc. Copyright 2002 Wiley-Liss, Inc. **Abstract:** *Background*: The majority of U.S. birth defects surveillance programs do not include elective terminations before 20 weeks gestation among the pregnancy outcomes covered. To assess the impact of defects among elective terminations before 20 weeks gestation, data from a birth defects registry that does include terminations before 20 weeks gestation were analyzed. *Methods*: Using information from the Texas Birth Defects Registry, the number of cases and rate of 49 conditions were

analyzed in two ways: excluding defects detected among elective pregnancy terminations before 20 weeks gestation, and including defects among terminations before 20 weeks. *Results*: By including defects detected among elective terminations before 20 weeks, the number of cases increased by five percent or greater for nine conditions: anencephaly (29%); spina bifida without anencephaly (13%); encephalocele (21%); Patau syndrome (19%); Edwards syndrome (11%); Down syndrome (6%); omphalocele (15%); gastroschisis (5%); and anophthalmia (7%). There was no impact for 27 conditions, for which there were no cases detected among elective terminations before 20 weeks. The greatest impact was observed for anencephaly; the rate of anencephaly increased from 2.76 to 3.56 per 10,000 live births when defects among elective terminations before 20 weeks were included. *Conclusions*: Excluding defects among elective terminations before 20 weeks results in counts and rates that are somewhat incomplete, especially for conditions that are more commonly detected and electively terminated before 20 weeks. The impact varies by condition.

Forrester MB, Canfield MA. 2000. **Evaluation of a system for linking birth defects registry records and vital records**. J Registry Management 27:93-97. Reprinted with permission. Copyright 2000, Journal of Registry Management by National Cancer Registrars Association, Alexandria, Virginia. **Abstract:** Linking to vital records may allow birth defects registries access to additional information on previously identified cases and an additional source of potential cases. The Texas Birth Defects Monitoring Division (TBDMD) evaluated a system for linking its records to the Texas Bureau of Vital Statistics birth and fetal death certificate records. Using six demographic variables, the TBDMD successfully linked 96.8% of its registry records to vital records. The rate was higher for live births (99.3%) than for fetal deaths (80.1%); some elective terminations (39.2%) were also linked to vital records. The majority of linked registry records (97.6%) demonstrated exact agreement among at least four of the six variables. Exact matches between registry and vital records data were highest for birth facility code (98.0%) and infant's date of birth (97.1%) and lowest for infant's last name (82.9%) and infant's first name (74.9%).

Nembhard WN, Waller DK, Sever LE, Canfield MA. 2001. **Patterns of First-Year Survival Among Infants with Selected Congenital Anomalies in Texas, 1995-1997**. Teratology 64:267-275. This material is used by permission of Wiley-Liss, Inc., a subsidiary of John Wiley & Sons, Inc. Copyright 2001 Wiley-Liss, Inc. **Abstract:** *Background*: Few registry-based studies have investigated survival among infants with congenital anomalies. We conducted a registry-based study to examine patterns and probability of survival during the first year of life among infants with selected congenital anomalies. *Methods*: Data from the Texas Birth Defects Monitoring Division were merged with linked birth-infant death files for 2,774 infants born January 1, 1995 to December 31, 1997, with at least 1 of 23 common anomalies. Deaths before the first birthday were assessed from infant death files. Kaplan-Meier was used to estimate first-year survival; first-year survival was assessed for specific anomalies and by the number of life-threatening anomalies. *Results*: Overall, 80.8% of infants with these 23 anomalies survived the first year of life. We observed the highest survival rates for infants with gastroschisis (92.9%, 95% CI = 86.8, 96.3), trisomy 21 (92.3%, 95% CI = 89.5, 94.4) or cleft lip with or without cleft palate (87.6%, 95% CI = 84.0, 90.5). Infants with intermediate survival rates included those with microcephaly (79.7%; 95% CI = 73.6, 84.6), tetralogy of Fallot (75.0%; 95% CI = 65.5, 82.2), or with diaphragmatic hernia (72.8%; 95% CI = 61.8, 81.2). As expected, all infants with anencephaly and almost all infants with trisomy 13 or trisomy 18 died during the first year of life. First-year survival declined as the number of co-occurring life-threatening anomalies increased. *Conclusions*: Overall, first-year survival for infants with congenital anomalies was high. Additional population-based studies are needed to quantify improvements in first-year survival.

Waller DK, Keddie AM, Canfield MA, Scheuerle AE. 2001. **Do infants with major congenital anomalies have an excess of macrosomia?** Teratology 64:311-317. This material is used by permission of Wiley-Liss, Inc., a subsidiary of John Wiley & Sons, Inc. Copyright 2001 Wiley-Liss, Inc. **Abstract:** *Background*: Infants that develop congenital anomalies may also have an excess prevalence of macrosomia (birth weight > or =4,000 g). This may indicate that abnormalities of glycemic control play a role in the etiology of birth defects. This study was undertaken to determine whether all infants with congenital anomalies have an excess of macrosomia and whether it is confined to specific types of anomalies. *Methods*: A case-control study was conducted, comparing the birth weights of 8,226 infants with congenital anomalies ascertained by the Texas Birth Defects Monitoring Division with those

of 965,965 infants without birth defects. Odds ratios were calculated to determine the association between birth weight and congenital anomalies, for 45 specific defects, and for all these defects combined. *Results*: For all 45 defects combined, a significant association occurred only in the highest birth weight category. Infants with congenital anomalies were more likely than infants without birth defects to have a birth weight > or =4,500 g (OR = 1.65; 95% CI = 1.39-1.96). Infants born with ventricular septal defects, atrial septal defects, ventricular hypertrophy, or anomalies of the great vessels were 1.5-2.5 times more likely to weigh > or =4,000 g than were infants without birth defects. Based on small numbers, a stronger excess of macrosomia was observed for infants with encephalocele, holoprosencephaly, anomalies of the corpus callosum, preaxial polydactyly, and omphalocele. *Conclusions*: Our data suggest that infants with specific congenital anomalies are more likely to be macrosomic than are infants without an anomaly. If these findings are confirmed, associations between macrosomia and specific types of birth defects may help to identify birth defects that are caused by alterations in glycemic control.

Waller DK, Keddie AM, Canfield MA, Scheuerle AE. 2002. **Sex ratios in infants with congenital anomalies**. Teratology 66:60. (Letter to the editor)

Waller DK, Pujazon MA, Canfield MA, Scheuerle AE, Byrne JLB. 2000. **Frequency of prenatal diagnosis of birth defects in Houston, Galveston, and the Lower Rio Grande Valley of Texas, 1995**. Fetal Diagnosis and Therapy, 15:348-354. Reprinted with permission from S. Karger AG, Basel. Copyright 2000, S. Karger AG, Basel. **Abstract:** *Background*: Estimates of the proportion of birth defects diagnosed before birth exist for only a few types of birth defects and for a few geographic regions in the United States. This population-based study examines rates of prenatal diagnosis for previously unstudied birth defects in a new geographic region. *Methods*: Active surveillance of 23 categories of birth defects among 111,902 infants born in 77 birthing hospitals in Texas in 1995 identified 852 infants or fetuses with major birth defects. Surveillance was conducted by the Texas Birth Defects Monitoring Program of the Texas Department of Health. Two regions were covered, the Houston/Galveston metropolitan area as well as the Lower Rio Grande Valley of Texas. Rates of prenatal diagnosis were evaluated for 23 different types of birth defects, using proportions and 95% confidence intervals. *Results*: One third of the 852 infants or fetuses with birth defects were prenatally diagnosed. Diagnosis rates varied greatly depending on the type of birth defects and were lower among infants born to Black and Hispanic women. More than 60% of anencephaly, encephalocele, gastroschisis and trisomies 13 and 18 were diagnosed antenatally. Many of the fetuses that were electively terminated had birth defects or combinations of birth defects that were potentially lethal. Prevalence rates for birth defects generally do not include fetuses that die or are electively terminated before 20 weeks of gestation. Thus, 36% of anencephaly, 21% of omphalocele, 15% of encephalocele and between 7 and 10% of spina bifida, hydrocephaly, renal agenesis, and trisomies 13, 18, and 21 were not included in our published rates. *Conclusions*: Published rates for specific types of birth defects are spuriously low. This should be considered when investigating alleged clusters and comparing rates of birth defects across geographic areas. Since many elective abortions are for lethal or potentially lethal birth defects, a major effect of prenatal diagnosis is the resultant decrease in infant mortality attributable to birth defects.

Williams LJ, Mai CT, Edmonds LD, Shaw GM, Kirby RS, Hobbs CA, Sever LE, Miller LA, Meaney FJ, Levitt M. 2002. **Prevalence of spina bifida and anencephaly during the transition to mandatory folic acid fortification in the United States**. Teratology 66:33-39. This material is used by permission of Wiley-Liss, Inc., a subsidiary of John Wiley & Sons, Inc. Copyright 2002 Wiley-Liss, Inc. **Abstract:** *Background*: In 1992, the United States Public Health Service recommended that all women of childbearing age consume 400 micrograms of folic acid daily. The Food and Drug Administration authorized the addition of synthetic folic acid to grain products in March 1996 with mandatory compliance by January 1998. The impact of these public health policies on the prevalence of neural tube defects needs to be evaluated. We sought to determine the prevalences of spina bifida and anencephaly during the transition to mandatory folic acid fortification. *Methods*: Twenty-four population-based surveillance systems were used to identify 5,630 cases of spina bifida and anencephaly from 1995-99. Cases were divided into three temporal categories depending on whether neural tube development occurred before folic acid fortification (January 1995 to December

1996), during optional fortification (January 1997 to September 1998), or during mandatory fortification (October 1998 to December 1999). Prevalences for each defect were calculated for each time period. Data were also stratified by programs that did and did not ascertain prenatally diagnosed cases. *Results*: The prevalence of spina bifida decreased 31% (prevalence ratio [PR] = 0.69, 95% confidence interval [CI] = 0.63-0.74) from the pre- to the mandatory fortification period and the prevalence of anencephaly decreased 16% (PR = 0.84, 95% CI = 0.75-0.95). Stratification by prenatal ascertainment did not alter results for spina bifida but did impact anencephaly trends. *Conclusions*: The decline in the prevalence of spina bifida was temporally associated with folic acid fortification of US grain supplies. The temporal association between fortification and the prevalence of anencephaly is unclear.

Yoon PW, Rasmussen SA, Lynberg MC, Moore CA, Anderka M, Carmichael SL, Costa P, Druschel C, Hobbs CA, Romitti PA, Langlois P, Edmonds LD, and The National Birth Defects Prevention Study. 2001. **The National Birth Defects Prevention Study**. Public Health Reports 116 Suppl 1:32-40. **Abstract:** The National Birth Defects Prevention Study was designed to identify infants with major birth defects and evaluate genetic and environmental factors associated with the occurrence of birth defects. The ongoing case-control study covers an annual birth population of 482,000 and includes cases identified from birth defect surveillance registries in eight states. Infants used as controls are randomly selected from birth certificates or birth hospital records. Mothers of case and control infants are interviewed and parents are asked to collect buccal cells from themselves and their infants for DNA testing. Information gathered from the interviews and the DNA specimens will be used to study independent genetic and environmental factors and gene-environment interactions for a broad range of birth defects. As of December 2000, 7,470 cases and 3,821 controls had been ascertained in the eight states. Interviews had been completed with 70% of the eligible case and control mothers, buccal cell collection had begun in all of the study sites, and researchers were developing analysis plans for the compiled data. This study is the largest and broadest collaborative effort ever conducted among the nation's leading birth defect researchers. The unprecedented statistical power that will result from this study will enable scientists to study the epidemiology of some rare birth defects for the first time. The compiled interview data and banked DNA of approximately 35 categories of birth defects will facilitate future research as new hypotheses and improved technologies emerge.

Manuscripts Under Review using Registry Data

Anderson JL, Waller DK, Shaw G, Watkins M, Werler M, Canfield MA. **Maternal obesity, gestational diabetes and central nervous system birth defects: A Texas case-control study.**

Conference Presentations and Posters using Registry Data

Canfield MA, Anderson JL, Waller DK, Palmer SE, Kaye CI. 2002. **Do women who have infants with neural tube defects recall and follow recommendations about folic acid for the prevention of a recurrence?** Presentation at the Texas Birth Defects Conference, March 7-8, 2002, Fort Worth, Texas.

Canfield MA, Suarez L. **Descriptive epidemiology of neural tube defects in a Texas Hispanic population**. Invited presentation at the Collaborative Meeting of the National Birth Defects Prevention Network and the International Clearinghouse for Birth Defects Monitoring Systems, September 20, 2002, Atlanta, Georgia. **Abstract:** Neural tube defects (anencephaly and spina bifida) have been of considerable public health concern to Texas since 1991, when a large cluster of anencephalic births occurred in the Hispanic border community of Brownsville, Texas. Data from the statewide Texas Birth Defects Registry from 1998-99 show that neural tube defects are more common among Hispanics than either whites or African Americans. Rates for spina bifida without anencephaly were 3.53, 4.79, and 3.38 per 10,000 live births for whites, Hispanics, and African Americans, respectively. Anencephaly rates were 2.79, 3.56, and 0.84 per 10,000 live births, respectively. Rates of spina bifida were particularly elevated among Hispanics living in the 14 Texas-Mexico border counties compared with non-border Hispanics and non-border whites (6.05 vs. 4.30 vs. 3.54 per 10,000 live births). Spina bifida rates were higher for Hispanic mothers with less than 7 years of education (5.12)

than for those with more than 12 years education (3.50). Hispanics in the youngest age category (<20 years) experienced the highest spina bifida rates, in contrast to the age pattern observed for whites and African Americans. Cases among Hispanics were somewhat less likely than white cases to result in an elective pregnancy termination (8% vs. 15%, respectively). The distribution of spina bifida cases by sex and level of lesion were similar for Texas Hispanics and whites. For anencephaly, we also observed an elevated rate among border county Hispanics (4.48 per 10,000 live births), relative to non-border Hispanics (3.20) and non-border whites (2.71). In contrast to spina bifida, anencephaly rates among Hispanics were roughly two-fold higher for Mexican- vs. American-born mothers (2.53 vs. 1.23, respectively). Texas anencephaly rates were markedly higher for Hispanic mothers with less than 7 years of education (2.69) vs. those with more than 12 years education (0.97). Compared with whites, pregnancy outcomes for Hispanic anencephaly cases were more likely to be live births and less likely to be late fetal deaths (20+ weeks). For example, live births comprised 33% of all pregnancy outcomes for anencephaly cases among Hispanics, but only 20% of cases among whites. These observations indicate that specific Hispanic subgroups would benefit from folic acid educational efforts.

Langlois P, Ethen MK. **Findings from the Texas Birth Defects Registry**. Presentation at the Texas Birth Defects Conference, March 7-8, 2002, Fort Worth, Texas. **Abstract:** Recent findings from the Texas Birth Defects Registry demonstrating the descriptive epidemiology of birth defects in Texas were presented. The distribution of birth defects was compared among groups defined by characteristics of person, such as maternal age, maternal race/ethnicity, and infant sex. The geographic distribution of birth defects was presented through analyses by Texas Public Health Region and county. Also included was a discussion of the limitations of data that are based on small numbers of cases.

Waller DK, Anderson JL, Nembhard W, Scheuerle AE, Wright D, Canfield MA. **Dieting, diet-related behaviors, and risk of neural tube defects: Results from the Texas Birth Defects Research Center, 1996 to 2000.** Poster presented at the National Birth Defects Prevention Network 4th Annual Meeting, January 29-31, 2001, San Antonio, Texas. **Abstract:** *Objective*: Our goal was to examine the associations between maternal dieting and diet-related behaviors in the periconceptional period and the risk of having offspring affected by neural tube defects. *Method*s: We analyzed data from a CDC-funded telephone interview of 112 mothers of infants and fetuses with neural tube defects (NTDs) from the Texas Birth Defects Registry and 226 mothers of control infants. *Results*: Women who reported dieting during the periconceptional period had no increase in the risk of NTDs (odds ratio 1.3, 95% CI 0.7-2.2). Mothers who reported they ate little or no food for a day or more during this period also had no increase in the risk of NTDs (odds ratio 1.4, 95% CI 0.8-2.5). For diet-related behaviors a preliminary analysis revealed a significant association between use of laxatives (odds ratio 3.0, 95% CI 1.2-7.4), pep pills (odds ratio 4.7, 95% CI 1.2-18.9), and vomiting (odds ratio 2.8, 95% CI 1.0-7.9) in the periconceptional period and an increased risk of having an infant with an NTD. A non-significant positive association was observed for use of diuretics (odds ratio 2.0, 95% CI 0.5-9.0). *Conclusions*: These results are preliminary and should therefore be interpreted cautiously. These diet-related behaviors may serve as markers for more severe diets, which may be associated with an increased risk of NTDs. They may also be associated with increased risk of an NTD, independently of dieting. We plan to conduct a more in-depth analysis of dieting and diet-related behaviors, and potential confounders that will include a larger number of cases and controls.

Graduate Student Research Projects using Registry Data

Anderson, James L. 2002. **The relationship of maternal body mass index prior to pregnancy, gestational diabetes, and congenital central nervous system malformations in the fetus and infant: A Texas case control study.** Doctor of Philosophy dissertation, The University of Texas Health Science Center at Houston, School of Public Health. Reprinted with permission of James L. Anderson. Copyright 2002, James L. Anderson. **Abstract:** Previous studies have consistently shown that obese women have an increased risk of having offspring with neural tube defects. Diabetes is also a strong and well-recognized risk factor for congenital anomalies. Obesity is a risk factor for diabetes, and both obesity and diabetes have some metabolic abnormalities in common, such as insulin resistance and hyperinsulinemia. It can be postulated that a common mechanism underlies the increased risk of

birth defects associated with diabetes and obesity. The objective of this case-control study was to evaluate maternal obesity (body mass index of greater than or equal to 30.0 kg/m^2) and the risk of having an offspring with one of the following congenital central nervous system (CNS) malformations: anencephaly, spina bifida, isolated hydrocephaly, holoprosencephaly, or encephalocele. We also evaluated whether risks associated with maternal obesity were modified by the presence of gestational diabetes. We used Texas telephone interview data from 491 case and 664 control mothers who gave birth between January 1997 and June 2001. Case mothers had offspring with one of the CNS birth defects, which were actively ascertained. Control mothers had non-malformed live births randomly selected from hospitals in areas of case ascertainment. Results were adjusted for maternal ethnicity, age, education, smoking, use of alcohol, and periconceptional multivitamin/folic acid use. Maternal obesity was associated with an increased risk of an offspring with anencephaly (OR 1.9, 95% CI 1.1-3.6), spina bifida (OR 2.4, 95% CI 1.5-3.8), or isolated hydrocephaly (OR 2.3, 95% CI 1.3-4.0); spina bifida and hydrocephaly showed a strong dose-response. The presence of gestational diabetes accentuated the effect of maternal obesity on spina bifida and holoprosencephaly. The association of maternal obesity and isolated hydrocephaly has not been demonstrated previously. If confirmed by other studies, hydrocephaly will be added to the list of congenital anomalies for which maternal obesity is a risk factor. The modification of the effects of obesity by gestational diabetes suggests obesity and diabetes may influence the risk of congenital anomalies by a common causal pathway. However, our estimates of effect modification were not sufficiently precise to exclude other explanations and our results should be interpreted cautiously.

Bell, Scott. 2002. **A Preliminary Investigation of Urban and Rural Disparities in the Rate of Birth Defects**. Master of Public Health student practicum project, Texas A&M University.

Fatunde, Adejoke Ariyike. 2001. **Association between oral clefts, prenatal alcohol and tobacco use in Public Health Regions 6 and 11 of Texas State**. Master of Public Health thesis, The University of Texas Health Science Center at Houston, School of Public Health.

Hashmi, Syed S. In Progress. **Prevalence of oral clefts in Texas: 1995-1999**. Master of Public Health thesis, The University of Texas Health Science Center at Houston, School of Public Health.

Keddie, Arlene. 2002. **Is low parental education associated with congenital heart defects?** Doctor of Philosophy dissertation, The University of Texas Health Science Center at Houston, School of Public Health. Reprinted with permission of Arlene Keddie. Copyright 2002, Arlene Keddie. **Abstract:** *Objective*: The purpose of this study was to determine if there are associations between low parental education and congenital heart defects. *Methods*: This was a cross-sectional study of 281,262 live born singletons, 1765 of whom were identified by the Texas Birth Defects Monitoring Division (TBDMD) as having heart defects without known chromosomal anomalies. Data on the specific diagnoses of these infants were linked to their corresponding birth certificates. Only infants born between January 1, 1995 and December 31, 1997, whose mothers resided in the Texas public health regions under surveillance by the TBDMD were included in the study. The number of years of schooling of the most educated parent was used to calculate crude, stratified and adjusted odds ratios. *Results*: An increase in the likelihood of having an infant with any type of congenital heart defect was found among parents with less than 16 years of education, compared to those with 16 or more years of schooling. The association became more marked with increasing paternal age, and was found among whites and Hispanics but not among blacks. Statistically significant associations with low parental education were found for ventricular septal defects, transposition of the great vessels and miscellaneous heart and vessel defects. Among whites, there was an inverse association between parental education and likelihood of having an infant with a severe ASD. This association was not found among non-whites. The suggestion of an association between low parental education and tetralogy of Fallot, was also found, but was not statistically significant. Parents with > 16 years of education had a greater likelihood of having an infant with severe endocardial cushion lesions or total anomalous pulmonary return than less well educated parents. *Conclusion*: This study suggests that parental education is associated with certain types of heart defects, especially among whites and Hispanics.

Other Publications, Presentations, Posters, and Completed Projects

Canfield MA, Case AP, Jayasuriya B, Dyer J, Wright D. 2002. **Texas Women and Folic Acid: An Update**. Presentation at the Texas Birth Defects Conference, March 7-8, 2002, Fort Worth, Texas. **Abstract:** Numerous studies have demonstrated that 400 micrograms of the vitamin folic acid as a daily supplement can prevent up to 75% of all cases of neural tube defects (anencephaly and spina bifida). This is the basis of recommendations by the Public Health Service, the National Academy of Sciences, and various health professional organizations for women of childbearing age to increase their folic acid intake through supplements and a varied diet. The Texas Women's Health Survey was conducted in both 1997 and 2001 with funds from the CDC-funded Texas Birth Defects Research Center, primarily to look at patterns of folic acid awareness and supplementation among Texas women of childbearing age (WCBA). Questions related to folic acid were identical to those in the March of Dimes/Gallup national survey conducted annually, so that Texas and national data could be compared. From 1997 to 2001, there was an increase in the general awareness about folic acid among WCBA; the percentage responding that they "had read or heard something about folic acid" was 66% in 1997 and 78% in 2001, essentially identical to national results. General awareness was substantially higher among whites and those with a college degree. When Texas women were asked what specifically they had read or heard about folic acid, the percentage responding accurately that it prevents birth defects was 16% in 1997 and 28% in 2001. The percentage responding accurately that folic acid should be taken before pregnancy was 14% in1997 and 25% in 2001. For this more specific folic acid knowledge, Texas women were more aware than those from the national sample. When asked where they had read or heard about folic acid, there was a marked increase in the percentage who mentioned health care providers (vs. print media or radio/television), from 24.8% in 1997 to 38.3% in 2001, and especially in Hispanics. Even with an increase in awareness about folic acid over the 4-year period, there was essentially no change in the percentage of WBCA who took folic acid containing supplements daily over the same time period (33% for both 1997 and 2001). In 2001, higher supplementation rates continued to be observed among non-Hispanic whites, (40%); residents of the Dallas-Ft. Worth region (41.2%) and non-border counties (34.6%); and those with a college degree (45.8%) and household incomes above $50,000 (47.6%). Among those who did not graduate from high school, there was no ethnic difference in folic acid supplementation. Women were roughly twice as likely to take folic acid-containing supplements daily if they knew about folic acid in either a general or specific sense.

Canfield MA, Ethen MK, Wright D, Dyer J. **Texas Women's Health Survey**. The Texas Women's Health Survey is an anonymous telephone survey conducted in 1997 and in 2001. Approximately 1200 women of childbearing ages (15-44) throughout Texas responded to each survey. The telephone interview collected information on women's knowledge and attitudes about preventing birth defects. Topics covered included use of vitamin supplements containing folic acid, knowledge about the risks of drinking alcohol during pregnancy, and attitudes concerning prenatal diagnostic services and pre-pregnancy planning.

Case AP, Canfield MA, Watkins M, Williams J, Bennett S, Przybyla S. **Consumption of highly fortified breakfast cereals among Texas women**. Poster presented at the CDC Conference on Birth Defects, Developmental Disabilities, and Disability and Health, September 17-19, 2002, Atlanta, Georgia. **Abstract:** *Objective*: Consumption of highly fortified cereal (HFC) provides 400 mcg of folic acid per serving, the amount recommended by the USPHS for prevention of neural tube defects among offspring of childbearing-age (CBA) women. However, little is known about consumption of HFC by CBA women, despite a substantial increase in the number of HFCs available in recent years. We assessed the consumption patterns of cereal and HFC among CBA women in Texas, as well as the impact of HFC products on the total consumption of synthetic folic acid. *Methods*: The Texas Women's Health Survey is a 15-minute computer-assisted telephone interview of 1200 Texas women of CBA conducted in 1997 and 2001, focusing on folic acid awareness and supplementation. The 2001 survey included two questions about breakfast cereal consumption in general, and about specific brand use. Interviewers were provided with a checklist of 44 private brand cereals known to contain 400 mcg of folic acid per serving, and recorded other brand responses. Frequency of breakfast cereal and HFC consumption were analyzed by various demographic characteristics. *Results*: 48.1% of women reported consuming some type of breakfast cereal at least twice a week (31.2% ate cereal 2-4

times/week and 16.9% ate cereal daily). This figure includes the 13.8% of women who reported consuming a HFC at least twice a week (9.2% ate a HFC 2-4 times/week and 4.6% ate a HFC daily). Roughly two-thirds of Texas women are not taking a supplement daily; in this subgroup, 3.5% ate HFC daily and 5.9% ate HFC 2-4 times/week. Demographic characteristics of cereal consumers and non-consumers will be provided. *Conclusions*: Consumption of HFC has the potential to increase the percentage of women who consume 400 mcg of folic acid daily, but present consumption levels are too low to make a major contribution. Cereals fortified with 100 mcg and 200 mcg per serving also affect folic acid consumption. More research is needed to define the contribution of HFC and other cereals to folic acid intake and blood folate levels and to identify appropriate strategies to promote cereal use among women with poor folate status.

Forrester M. **Etiology and Epidemiology of Down Syndrome**. Presentation at the Texas Birth Defects Conference, March 7-8, 2002, Fort Worth, Texas. **Abstract:** Down syndrome is caused by excess genetic material from chromosome 21, usually when a complete extra chromosome 21 is present (trisomy 21). Trisomy 21 results from nondisjunction where one cell receives and extra copy of chromosome 21. This extra chromosome 21 is most often of maternal origin and often results from nondisjunction in the first stage of meiosis. Problems when examining the epidemiology of Down syndrome are that not all potential cases are karyotyped or the karyotype is not always obtained, the majority of Down syndrome conceptuses result in fetal death, and a portion of conceptuses are electively terminated. Down syndrome prevalence depends on the manner in which it is calculated, thus limiting comparison between populations. The most well known risk factor for Down syndrome is advanced maternal age. Secular trends and racial/ethnic differences in Down syndrome rates have been observed. Down syndrome is more common among male live births than female live births. Down syndrome has not been associated with maternal hypothyroidism, hyperthyroidism, the common cold, epilepsy, hypertension, diabetes, or tobacco smoking. Down syndrome risk is reduced with maternal coffee and alcohol use during pregnancy, possibly because these exposures reduce the viability of Down syndrome conceptuses. Recently, Down syndrome risk has been linked to mutations in folate metabolism genes.

Kirby RS, Canfield MA. 2002. **Surveillance methods, public health applications, and epidemiologic research using population-based birth defects registries**. Teratology 66:S1-S2. This article precedes and introduces the reader to the National Birth Defects Prevention Network 2002 Annual Report, which is included in the same issue of the journal. The articles contained in the report are briefly described and the implications of each are pointed out.

Langlois P, Lynberg M. 2001. **Tap water disinfection byproducts and birth defects**. Disease Prevention News 61(23):1-4. Disinfection of public drinking water has been one of the greatest public health successes in the last century. However, chlorine disinfectants can react with organic substances in the water to create disinfection byproducts such as trihalomethanes (THMs), which include chloroform, bromodichloromethane, dibromochloromethane, and bromoform and haloacetic acids (HAAs). This report summarizes studies dealing with disinfection byproducts and birth defects. The full text of this report is available at www.tdh.state.tx.us/phpep/dpn/ISSUES/dpn61n23.pdf.

Lynberg M, Nuckols JR, Langlois P, Ashley D, Singer P, Mendola P, Wilkes C, Krapfl H, Miles E, Speight V, Lin B, Small L, Miles A, Bonin M, Zeitz P, Tadkod A, Henry J, Forrester MB. 2001. **Assessing exposure to disinfection by-products in women of reproductive age living in Corpus Christi, Texas, and Cobb county, Georgia: descriptive results and methods**. Environmental Health Perspectives 109:597-604. **Abstract:** We conducted a field study in Corpus Christi, Texas, and Cobb County, Georgia, to evaluate exposure measures for disinfection by-products, with special emphasis on trihalomethanes (THMs). Participants were mothers living in either geographic area who had given birth to healthy infants from June 1998 through May 1999. We assessed exposure by sampling blood and water and obtaining information about water use habits and tap water characteristics. Two 10-mL whole blood samples were collected from each participant before and immediately after her shower. Levels of individual THM species (chloroform, bromodichloromethane, dibromochloromethane, and bromoform) were measured in whole blood [parts per trillion (pptr)] and in water samples (parts per billion). In the Corpus Christi water samples, brominated compounds accounted for 71% of the total

THM concentration by weight; in Cobb County, chloroform accounted for 88%. Significant differences in blood THM levels were observed between study locations. For example, the median baseline blood level of bromoform was 0.3 pptr and 3.5 pptr for participants in Cobb County and Corpus Christi, respectively ($p = 0.0001$). Differences were most striking in blood obtained after showering. For bromoform, the median blood levels were 0.5 pptr and 17 pptr for participants in Cobb County and Corpus Christi, respectively ($p = 0.0001$). These results suggest that blood levels of THM species vary substantially across populations, depending on both water quality characteristics and water use activities. Such variation has important implications for epidemiologic studies of the potential health effects of disinfection by-products.

Miles AM, Singer PC, Ashley DL, Lynberg MC, Mendola P, Langlois PH, Nuckols JR. 2002. **Comparison of trihalomethanes in tap water and blood**. Environmental Science and Technology 36:1692-1698. Reprinted with permission from Environmental Science and Technology. Copyright 2002, American Chemical Society. **Abstract:** Trihalomethane (THM) concentrations in blood and tap water were measured for 50 women living in two locations with different bromide concentrations and disinfectant types. Blood samples were taken from each woman early in the morning prior to any major water-use activity and again immediately after showering. Each residence was sampled for THMs in tap water prior to the woman's shower. Cobb County, GA, tap water exhibited high THM concentrations composed primarily of chloroform. Corpus Christi, TX, tap water exhibited lower THM concentrations with significant proportions of brominated THMs. THMs in tap water and blood were compared using mole fraction speciation, extent of bromine incorporation, and correlation analysis. Results indicated that THMs in the blood rose significantly as a result of showering, that showering shifted the THM distribution in the blood toward that found in the corresponding tap water, and that THMs measured in the blood of women living in the two locations reflected species and concentration differences in their respective tap waters. In general, blood concentrations were not significantly correlated with tap water concentrations. This finding suggests that other factors, in addition to tap water concentrations, may be important in determining THM concentrations in the blood.

Scheuerle AE, Wright D. **Ethical Issues Encountered in the Establishment of the Texas Birth Defects Research Center**. Poster presented at the American College of Medical Genetics Annual Meeting, March 9-12, 2000, Palm Springs, California.

Sever L, Canfield MA, Tinkle M. **The Neural Tube Defect (NTD) Occurrence Prevention Project**. The objectives of this project were to evaluate and increase awareness among Texas providers about folic acid and prevention of neural tube defects, and to establish this prevention message as a standard of care for all women of childbearing age.

Strahan JE, Canfield MA, Drummond-Borg, LM, Neill SU. 2002. **Ethnic and gender patterns for five congenital disorders in Texas from 1992 through 1998**. Texas Medicine 98(9):80-86. Reprinted with permission of Texas Medicine and the Texas Medical Association. Copyright 2002, Texas Medicine. **Abstract:** The Texas Department of Health's Newborn Screening Program screens for five inherited disorders: phenylketonuria (PKU), congenital adrenal hyperplasia (CAH), congenital hypothyroidism (CH), galactosemia (GAL), and sickle cell disease (SCD). The objective of this study was to determine the prevalence of these disorders and to describe ethnic and gender patterns in their distribution. Cases were identified from blood specimens collected at birth from live births in Texas from 1992 through 1998. During this time, the overall prevalence of these disorders per 10,000 live births was 0.70 for PKU, 0.21 for GAL, 4.18 for CH, 1.03 for CAH, and 3.92 for SCD. Ethnic and gender disparities were observed among PKU, CH, CAH, and SCD prevalence. Results suggest that unidentified mutations and environmental factors may exist that contribute to these patterns. This warrants further investigation to determine possible modifiable risk factors for populations with higher prevalence.

Wright D, Kamen B, Smith A, Novoa A. **Blood Folate Survey of Texas Women, A Feasibility Study**. Venous blood (arm draw) specimens and capillary blood (finger-stick dropped on blood spots cards) were collected from 600 self-selecting, non-pregnant women residing in Central Texas, East Texas, and South Texas localities. Subjects self-administered a 10 minute computer-assisted questionnaire regarding their awareness and consumption of folic acid-containing supplements and foods, and use

of over-the-counter and prescription medications that are known folate-antagonists. Laboratory testing included assaying blood specimens for folate levels and comparing the stability of dried blood folate to venous blood source of folate.

Other Ongoing Projects

Canfield MA, Case AP. **Folic Acid Awareness Module of the Texas Behavioral Risk Factor Surveillance Survey**. The Texas Behavioral Risk Factor Surveillance System (BRFSS), initiated in 1987, is a federally funded telephone survey conducted on a monthly basis with 1500 randomly selected adult Texans to collect data on lifestyle risk factors contributing to the leading causes of death and chronic diseases. As part of the national BRFSS, Texas can select from a list of standardized questions, known as optional modules, which are asked of a specific target population. From 1999 through 2001, the Texas Birth Defects Research Center (TBDRC) funded the use of a folic acid module. This module includes standard questions determined by the Centers for Disease Control and Prevention, plus the following two questions written by the TBDRC: "Has a doctor or nurse ever advised you to take multivitamins or supplements?" and "What would you say is the main reason that you do not take any vitamin pills or supplements?" Educational attainment was strongly correlated with a woman's awareness of folic acid and birth defects, daily folic acid supplementation, and whether a health care provider ever advised her to take a vitamin or supplement. Although the survey provided a choice of four pre-written answers to the second question above, more than half gave another answer or responded "Don't know". Analysis of these reasons given will provide a rich resource for designing folic acid awareness strategies in Texas.

Canfield MA, PhD, Trevino J, Paz E, Dutton RJ. **Texas-Mexico Border States Birth Defects Project**. Key birth defects contacts in Texas and the four bordering Mexican states of Tamaulipas, Coahuila, Chihuaha, and Nuevo Leon have met four times over the last several years to enhance communication, data exchange, surveillance, and prevention of birth defects.

Canfield MA, PhD, Waller DK, Anderson JL, Finnell R, Scheuerle AE. **Hispanic Origin, Maternal Obesity, and Central Nervous System Defects**. This study examines the metabolic-genetic-environmental risks associated with certain birth defects of the brain and spinal cord. The study consists of a telephone interview with 400 mothers of infants affected by anencephaly, spina bifida, isolated hydrocephaly, or holoprosencephaly, and with 400 mothers of control infants.

Shapira S, Ornelas E, Enciso V, Canfield MA. **The Neural Tube Defect (NTD) Recurrence Prevention Project**. This project is an educational intervention for families with infants affected by neural tube defects. Its purpose is to promote to mothers and health care providers the pre-conception consumption of high-dose folic acid to reduce the risk of subsequent NTD-affected pregnancies. Implementation is in progress in San Antonio, Harlingen, El Paso, and Laredo.